



**Connecticut Department of Public Health**

**Testimony Presented before the Public Health Committee  
February 28, 2014**

**Commissioner Jewel Mullen, MD, MPH, MPA  
860-509-7101**

**House Bill No. 5147: An Act Concerning Newborn Screening for Globoid Cell Leukodystrophy and Cytomegalovirus and Establishing a Public Education Program for Cytomegalovirus**

The Department of Public Health (DPH) would like to provide the following information regarding House Bill No. 5147.

HB 5147 would require screening for globoid cell leukodystrophy, also known as Krabbe disease, as part of the Newborn Screening (NBS) panel tested by the Public Health Laboratory (PHL). The bill would also require that any baby who fails a newborn hearing test be screened for cytomegalovirus (CMV), but we believe the legislative intent is that this screening would be a hospital mandate and would occur in the clinical care setting. It should be noted that the PHL only receives blood spots for NBS, not urine or saliva which are the standard matrices for CMV testing.

Krabbe disease is a genetic disorder caused by an enzyme deficiency which affects nervous system development. In the infantile form, symptom onset is typically before one year of age, and includes muscular weakness, feeding difficulties, vision loss, seizures, and slowed mental and physical development. As the muscular wasting progresses, the infant's ability to move, chew, swallow, and breathe lessens. The infantile form of the disease is generally fatal before age 2; incidence in the United States is approximately 1:100,000. Although we understand the interest in addressing this devastating disease, the available science is unfortunately not yet in place to support universal newborn screening.

The Secretary's Advisory Committee on Heritable Disorders in Newborns and Children, within the federal Department of Health and Human Services, was petitioned to include Krabbe disease in the recommended uniform screening panel. Following a careful review of available evidence, the Committee concluded that, while there could be a benefit from early identification and intervention, substantial harm is also possible, either from testing/identification, from treatment/other interventions, or both. The Committee determined not to recommend adding the condition to the recommended panel, noting that

*Phone: (860) 509-7269, Fax: (860) 509-7100, Telephone Device for the Deaf (860) 509-7191  
410 Capital Avenue - MS # 13GRE, P.O. Box 340308 Hartford, CT 06134  
An Equal Opportunity Employer*

there are significant gaps in evidence that must be addressed before further consideration of its inclusion. Treatment for Krabbe disease requires stem cell transplantation and carries significant risks, which must be weighed against the current difficulty predicting which babies will be affected by the severe infantile form of the disease.

A very small minority of states are offering Krabbe testing within their NBS panel. Their experience is that screening is problematic for a variety of reasons, including the generation of high numbers of false positive results that require further confirmatory testing. This significantly increases the cost of screening, tracking, and follow-up testing. The situation is further complicated by the identification of infants with mutations of unknown clinical significance.

Should this screening be mandated, the responsibility could not be conducted without additional resources for instrumentation, reagents, and staff for the PHL.

Thank you for your consideration of the Department's views on this bill.